



SEQUENCE LISTING

<110> Daniel H. Cohn
Muhammad Faiyaz ul Haque
Lily M. King
Deborah Krakow

<120> GENETIC MARKER FOR
SPONDYLOEPIMETAPHYSEAL DYSPLASIA

<130> 18810-81553

<140> US 09/898,200
<141> 2001-07-02

<150> 09/399,212
<151> 1999-09-17

<160> 33

<170> FastSEQ for Windows Version 4.0

<210> 1
<211> 2014
<212> DNA
<213> Homo sapiens

<400> 1

ctgctgccgc	cggccgcgc	gccgtccctg	cgtccttcgg	tctctgtcc	cgggaccgg	60
ctccggcgca	gccagccagc	atgtcgggga	tcaagaagca	aaagacggag	aaccaggaga	120
aatccaccaa	tgttagtctat	caggcccacc	atgtgagcag	gaataagaga	gggcaagtgg	180
ttggaacaag	gggtgggttc	cgaggatgta	ccgtgtggct	aacaggctc	tctggtgctg	240
gaaaaacaac	gataagttt	gccctggagg	agtaccttgt	ctcccatgccc	atcccttgtt	300
actccctgga	tggggacaat	gtccgtcatg	gccttaacag	aatctcgga	ttctctcctg	360
gggacagaga	ggaaaatatac	cgccggattt	ctgaggtggc	taagctttt	gctgatgctg	420
gtctggctcg	cattaccaggc	tttatttctc	cattcgcaaa	ggatcgtgag	aatgcccga	480
aaatacatga	atcagcaggg	ctgccattct	ttgaaatatt	tgttagatgca	cctctaaata	540
tttgtgaaag	cagagacgta	aaaggctct	ataaaagggc	cagagctggg	gagattaaag	600
gatttacagg	tattgattct	gattatgaga	aacctgaaac	tcctgagcgt	gtgcttaaaa	660
ccaatttgtc	cacagttagt	gactgtgtcc	accaggttagt	ggaacttctg	caagagcaga	720
acattgtacc	ctatactata	atcaaagata	tccacgaact	ctttgtccg	gaaaacaaac	780
ttgaccacgt	ccgagcttag	gctgaaactc	tcccttcatt	atcaattact	aagctggatc	840
tccagtgggt	ccaggtttt	agcgaaggct	ggccactcc	cctcaaagg	ttcatgcggg	900
agaaggagta	cttacaggtt	atgcacttt	acaccctgct	agatgatggc	gtatcaaca	960
tgagcatccc	cattgtactg	cccgctctg	cagaggataa	gacacggctg	gaagggtgca	1020
gcaagtttgt	cctggcacat	ggtggacgga	ggtagctat	cttacgagac	gctgaattct	1080
atgaacacag	aaaagagggaa	cgctgttccc	gtgtttgggg	gacaacatgt	acaaaacacc	1140
cccatatcaa	aatgggtatg	gaaagtgggg	actggcttgt	tggtggagac	cttcagggtgc	1200
tggagaaaat	aagatggaaat	gatgggctgg	accaataccg	tctgacacct	ctggagctca	1260
aacagaaatg	taaagaaatg	aatgctgatg	cggtgtttgc	attccagttt	cgcaatcctg	1320
tccacaatgg	ccatgccctg	ttgatgcagg	acacctgccg	caggctccta	gagagggct	1380
acaagcaccc	ggtcctccta	ctacaccctc	tggcggtcg	gaccaaggat	gacgatgtgc	1440
ctctagactg	gcggatgaag	cagcacgcgg	ctgtgctcga	ggaagggtc	ctggatccca	1500
agtcaaccat	tgttgccatc	tttccgtctc	ccatgttata	tgctggcccc	acagaggtcc	1560

agtggcactg caggtcccg atgattgcgg gtgccaattt ctacattgtg gggagggacc 1620
ctcgaggaat gccccatctt gaaaccaaga aggatctgtt tgaaccact catggggca 1680
aggctttagt catggccctt ggctcacct ctgtggaaat cattccattt cgagtggctg 1740
cctacaacaa agccaaaaaa gccatggact tctatgtcc agcaaggcac aatgagttt 1800
acttcatctc aggaactcga atgaggaagc tcgcccggga aggagagaat cccccagatg 1860
gcttcatggc ccccaaagca tggaaaggcc tgacagatata ttacagggtcc ctggagaaga 1920
actaagcctt tgggtccaga gtttctttt gaagtgcctt ttgattaccc tttcttatttt 1980
tatgattaga tgcttgat taaattgtt ctca 2014

<210> 2

<211> 2000

<212> DNA

<213> Mus musculus

<400> 2

gtattctcaa catcagatat catgtcttgg aggaagttac ctaaactctg aagaatttac 60
atgtctgcaa attcaaaaat gaaccataaa agagaccagc aaaaatccac caatgtggc 120
taccaggccc atcatgttag caggaacaag agaggacaag tgggtggac caggggagga 180
ttccgaggat gtaccgtgt gctaacaggt ctctctgtt ctggaaaac aaccataagc 240
tttgctttgg aagagtacct tttatctcac gccatcccat gttactccct ggatggggac 300
aatgtccgtc atggccttaa taagaacctg ggattctctg ccggggaccg agaagagaat 360
atccgcccgg a tcgcccggat ggccaagctc tttgccgacg ccggcctggg ttgcattcacc 420
agctttatct ctcctttgc aaaggatcgt gagaatccc gaaaaatcca cgaatcagca 480
ggactccgt tctttgagat cttttagat ggcctttaa atatctgtt aagccgagac 540
gtaaaaggac tctacaaaacg agcccgagca ggagagatta aagggtttac aggcatcgat 600
tctgactatg agaaacactga aactccagag tttgtgttca agaccaactt gtcttcagta 660
agcgactgtg tgcaacaggt ggtggactt ttgcaggagc agaacattgtt accccacacc 720
accatcaaag gcatccacga actctttgtt ccagaaaaca aagtcgtatca aatccgagct 780
gaggcagaga ctctccatc actaccaatt accaagctgg atctgcagtg ggtgcagatt 840
ctgagtgaag gctggccac tcccctcaaa ggctttatgc gggagaagga atacttgca 900
actctacact tcgacactct actggacgtt ggagtcatca acatgagttt tcccattgtt 960
ttgcccgtt ctgcggatga caaggcacgg ctcgaagggt gcagcaaatt tgccttgatg 1020
tacgaagggtc ggagggtcgc tctattacag gaccctgaat tctatgagca taggaaagag 1080
gagcgttggt ctcgtgtgtt gggacagcc actgcaaaacg acccccattt caaaatggg 1140
atggaaaagt gggactggct tttgggttca gacctacagg tgcttagagag aataagggtgg 1200
gacgatgggc tggaccaata ccgccttacg cctctggac tcaaacagaa gtgtaaagac 1260
atgaatgctg atgccgttt tgcatccatc ttgcgcatac ctgtccacaa tggcatgcc 1320
ctcctgtatgc aggacacccg ccgcaggctc ctggagaggg gttacaagca cccagtcctc 1380
ctgctccacc ctcttgggg ctggaccaag gacgatgtacg tacctcttga atggaggatg 1440
aaacagcatg cagctgtact ggaggaaagg gtcctggatc ccaagtcaac tattttgttgc 1500
atcttccat ctcctatgtt atacgttgtt cccacagagg tccagtggca ttgcagatgc 1560
cgatgattt caggagccaa tttctacatt gtgggttaggg atcccgagg aatgccccat 1620
cctgagacaa agaaagaccc atatgaaccc acccacgggg gcaaggctt gagtatggcc 1680
cctggccttta cctctgttca aataattccg ttccgagggtt ctgcctacaa taaaattaaa 1740
aaggccatgg acttttatgtt tccagcaagg cacgaggatg ttgacttcat ctcaggaact 1800
cgcatgagga agctcgcccg ggaaggagaa gatccccag atggcttcat ggccccgaaa 1860
gcgtggaaag ttttgcacat tttactacagg tctctggaga agaccaacta ggtgctcctg 1920
gctctggctt ctccctcaag tgctctgtt ccattttttt tttcttatttt tttttttttt 1980
ctgctctgtt tccaaattgtt 2000

<210> 3

<211> 20

<212> DNA

<213> Homo sapiens

<400> 3		
tggaccaagg atgacgtatgt		20
<210> 4		
<211> 20		
<212> DNA		
<213> Homo sapiens		
<400> 4		
cggaaagatg gcaacaatgg		20
<210> 5		
<211> 20		
<212> DNA		
<213> Homo sapiens		
<400> 5		
ctggtgctgg aaaaacaacg		20
<210> 6		
<211> 22		
<212> DNA		
<213> Homo sapiens		
<400> 6		
tgcgaatgga gaaataaagc tg		22
<210> 7		
<211> 615		
<212> PRT		
<213> Homo sapiens		
<400> 7		
Met Ser Gly Ile Lys Lys Gln Lys Thr Glu Asn Gln Gln Lys Ser Thr		
1 5 10 15		
Asn Val Val Tyr Gln Ala His His Val Ser Arg Asn Lys Arg Gly Gln		
20 25 30		
Val Val Gly Thr Arg Gly Gly Phe Arg Gly Cys Thr Val Trp Leu Thr		
35 40 45		
Gly Leu Ser Gly Ala Gly Lys Thr Thr Ile Ser Phe Ala Leu Glu Glu		
50 55 60		
Tyr Leu Val Ser His Ala Ile Pro Cys Tyr Ser Leu Asp Gly Asp Asn		
65 70 75 80		
Val Arg His Gly Leu Asn Arg Asn Leu Gly Phe Ser Pro Gly Asp Arg		
85 90 95		
Glu Glu Asn Ile Arg Arg Ile Ala Glu Val Ala Lys Leu Phe Ala Asp		
100 105 110		
Ala Gly Leu Val Cys Ile Thr Ser Phe Ile Ser Pro Phe Ala Lys Asp		
115 120 125		
Arg Glu Asn Ala Arg Lys Ile His Glu Ser Ala Gly Leu Pro Phe Phe		
130 135 140		
Glu Ile Phe Val Asp Ala Pro Leu Asn Ile Cys Glu Ser Arg Asp Val		
145 150 155 160		
Lys Gly Leu Tyr Lys Arg Ala Arg Ala Gly Glu Ile Lys Gly Phe Thr		
165 170 175		

Gly Ile Asp Ser Asp Tyr Glu Lys Pro Glu Thr Pro Glu Arg Val Leu
 180 185 190
 Lys Thr Asn Leu Ser Thr Val Ser Asp Cys Val His Gln Val Val Glu
 195 200 205
 Leu Leu Gln Glu Gln Asn Ile Val Pro Tyr Thr Ile Ile Lys Asp Ile
 210 215 220
 His Glu Leu Phe Val Pro Glu Asn Lys Leu Asp His Val Arg Ala Glu
 225 230 235 240
 Ala Glu Thr Leu Pro Ser Leu Ser Ile Thr Lys Leu Asp Leu Gln Trp
 245 250 255
 Val Gln Val Leu Ser Glu Gly Trp Ala Thr Pro Leu Lys Gly Phe Met
 260 265 270
 Arg Glu Lys Glu Tyr Leu Gln Val Met His Phe Asp Thr Leu Leu Asp
 275 280 285
 Asp Gly Val Ile Asn Met Ser Ile Pro Ile Val Leu Pro Val Ser Ala
 290 295 300
 Glu Asp Lys Thr Arg Leu Glu Gly Cys Ser Lys Phe Val Leu Ala His
 305 310 315 320
 Gly Gly Arg Arg Val Ala Ile Leu Arg Asp Ala Glu Phe Tyr Glu His
 325 330 335
 Arg Lys Glu Glu Arg Cys Ser Arg Val Trp Gly Thr Thr Cys Thr Lys
 340 345 350
 His Pro His Ile Lys Met Val Met Glu Ser Gly Asp Trp Leu Val Gly
 355 360 365
 Gly Asp Leu Gln Val Leu Glu Lys Ile Arg Trp Asn Asp Gly Leu Asp
 370 375 380
 Gln Tyr Arg Leu Thr Pro Leu Glu Leu Lys Gln Lys Cys Lys Glu Met
 385 390 395 400
 Asn Ala Asp Ala Val Phe Ala Phe Gln Leu Arg Asn Pro Val His Asn
 405 410 415
 Gly His Ala Leu Leu Met Gln Asp Thr Cys Arg Arg Leu Leu Glu Arg
 420 425 430
 Gly Tyr Lys His Pro Val Leu Leu His Pro Leu Gly Gly Trp Thr
 435 440 445
 Lys Asp Asp Asp Val Pro Leu Asp Trp Arg Met Lys Gln His Ala Ala
 450 455 460
 Val Leu Glu Glu Gly Val Leu Asp Pro Lys Ser Thr Ile Val Ala Ile
 465 470 475 480
 Phe Pro Ser Pro Met Leu Tyr Ala Gly Pro Thr Glu Val Gln Trp His
 485 490 495
 Cys Arg Ser Arg Met Ile Ala Gly Ala Asn Phe Tyr Ile Val Gly Arg
 500 505 510
 Asp Pro Ala Gly Met Pro His Pro Glu Thr Lys Lys Asp Leu Tyr Glu
 515 520 525
 Pro Thr His Gly Gly Lys Val Leu Ser Met Ala Pro Gly Leu Thr Ser
 530 535 540
 Val Glu Ile Ile Pro Phe Arg Val Ala Ala Tyr Asn Lys Ala Lys Lys
 545 550 555 560
 Ala Met Asp Phe Tyr Asp Pro Ala Arg His Asn Glu Phe Asp Phe Ile
 565 570 575
 Ser Gly Thr Arg Met Arg Lys Leu Ala Arg Glu Gly Glu Asn Pro Pro
 580 585 590
 Asp Gly Phe Met Ala Pro Lys Ala Trp Lys Val Leu Thr Asp Tyr Tyr
 595 600 605
 Arg Ser Glu Met Asp Lys Asn

<210> 8
<211> 617
<212> PRT
<213> Mus musculus

<400> 8

Met	Ser	Ala	Asn	Phe	Lys	Met	Asn	His	Lys	Arg	Asp	Gln	Gln	Lys	Ser	
1				5				10						15		
Thr	Asn	Val	Val	Tyr	Gln	Ala	His	His	Val	Ser	Arg	Asn	Lys	Arg	Gly	
				20				25						30		
Gln	Val	Val	Gly	Thr	Arg	Gly	Gly	Phe	Arg	Gly	Cys	Thr	Val	Trp	Leu	
				35				40						45		
Thr	Gly	Leu	Ser	Gly	Ala	Gly	Lys	Thr	Thr	Ile	Ser	Phe	Ala	Leu	Glu	
					50			55						60		
Glu	Tyr	Leu	Val	Ser	His	Ala	Ile	Pro	Cys	Tyr	Ser	Leu	Asp	Gly	Asp	
				65			70			75				80		
Asn	Val	Arg	His	Gly	Leu	Asn	Lys	Asn	Leu	Gly	Phe	Ser	Ala	Gly	Asp	
					85				90					95		
Arg	Glu	Glu	Asn	Ile	Arg	Arg	Ile	Ala	Glu	Val	Ala	Lys	Leu	Phe	Ala	
				100				105						110		
Asp	Ala	Gly	Leu	Val	Cys	Ile	Thr	Ser	Phe	Ile	Ser	Pro	Phe	Ala	Lys	
				115				120						125		
Asp	Arg	Glu	Asn	Ala	Arg	Lys	Ile	His	Glu	Ser	Ala	Gly	Leu	Pro	Phe	
				130			135							140		
Phe	Glu	Ile	Phe	Val	Asp	Ala	Pro	Leu	Asn	Ile	Cys	Glu	Ser	Arg	Asp	
				145			150			155				160		
Val	Lys	Gly	Leu	Tyr	Lys	Arg	Ala	Arg	Ala	Gly	Glu	Ile	Lys	Gly	Phe	
					165				170					175		
Thr	Gly	Ile	Asp	Ser	Asp	Tyr	Glu	Lys	Pro	Glu	Thr	Pro	Glu	Cys	Val	
				180				185						190		
Leu	Lys	Thr	Asn	Leu	Ser	Ser	Val	Ser	Asp	Cys	Val	Gln	Gln	Val	Val	
				195				200						205		
Glu	Leu	Leu	Gln	Glu	Gln	Asn	Ile	Val	Pro	His	Thr	Thr	Ile	Lys	Gly	
				210			215				220					
Ile	His	Glu	Leu	Phe	Val	Pro	Glu	Asn	Lys	Val	Asp	Gln	Ile	Arg	Ala	
				225			230			235				240		
Glu	Ala	Glu	Thr	Leu	Pro	Ser	Leu	Pro	Ile	Thr	Lys	Leu	Asp	Leu	Gln	
				245				250						255		
Trp	Val	Gln	Ile	Leu	Ser	Glu	Gly	Trp	Ala	Thr	Pro	Leu	Lys	Gly	Phe	
				260				265						270		
Met	Arg	Glu	Lys	Glu	Tyr	Leu	Gln	Thr	Leu	His	Phe	Asp	Thr	Leu	Leu	
				275				280				285				
Asp	Asp	Gly	Val	Ile	Asn	Met	Ser	Ile	Pro	Ile	Val	Leu	Pro	Val	Ser	
				290			295			300						
Ala	Asp	Asp	Lys	Ala	Arg	Leu	Glu	Gly	Cys	Ser	Lys	Phe	Ala	Leu	Met	
				305			310			315				320		
Tyr	Glu	Gly	Arg	Arg	Val	Ala	Leu	Leu	Gln	Asp	Pro	Glu	Phe	Tyr	Glu	
					325				330					335		
His	Arg	Lys	Glu	Glu	Arg	Cys	Ser	Arg	Val	Trp	Gly	Thr	Ala	Thr	Ala	
					340			345						350		
Lys	His	His	Pro	His	Ile	Lys	Met	Val	Met	Glu	Ser	Gly	Asp	Trp	Leu	Val
					355			360						365		

Gly Gly Asp Leu Gln Val Leu Glu Arg Ile Arg Trp Asp Asp Gly Leu
 370 375 380
 Asp Gln Tyr Arg Leu Thr Pro Leu Glu Leu Lys Gln Lys Cys Lys Asp
 385 390 395 400
 Met Asn Ala Asp Ala Val Phe Ala Phe Gln Leu Arg Asn Pro Val His
 405 410 415
 Asn Gly His Ala Leu Leu Met Gln Asp Thr Arg Arg Arg Leu Leu Glu
 420 425 430
 Arg Gly Tyr Lys His Pro Val Leu Leu His Pro Leu Gly Gly Trp
 435 440 445
 Thr Lys Asp Asp Asp Val Pro Leu Glu Trp Arg Met Lys Gln His Ala
 450 455 460
 Ala Val Leu Glu Glu Arg Val Leu Asp Pro Lys Ser Thr Ile Val Ala
 465 470 475 480
 Ile Phe Pro Ser Pro Met Leu Tyr Ala Gly Pro Thr Glu Val Gln Trp
 485 490 495
 His Cys Arg Cys Arg Met Ile Ala Gly Ala Asn Phe Tyr Ile Val Gly
 500 505 510
 Arg Asp Pro Ala Gly Met Pro His Pro Glu Thr Lys Lys Asp Leu Tyr
 515 520 525
 Glu Pro Thr His Gly Gly Lys Val Leu Ser Met Ala Pro Gly Leu Thr
 530 535 540
 Ser Val Glu Ile Ile Pro Phe Arg Val Ala Ala Tyr Asn Lys Ile Lys
 545 550 555 560
 Lys Ala Met Asp Phe Tyr Asp Pro Ala Arg His Glu Glu Phe Asp Phe
 565 570 575
 Ile Ser Gly Thr Arg Met Arg Lys Leu Ala Arg Glu Gly Glu Asp Pro
 580 585 590
 Pro Asp Gly Phe Met Ala Pro Lys Ala Trp Lys Val Leu Thr Asp Tyr
 595 600 605
 Tyr Arg Ser Glu Met Asp Lys Thr Asn
 610 615

<210> 9
 <211> 1845
 <212> DNA
 <213> Homo sapiens

<400> 9
 atgtcgggga tcaagaagca aaagacggag aaccagcaga aatccaccaa tgtagtctat 60
 caggcccacc atgtgaggcag gaataagaga gggcaagtgg ttggaacaaag ggggtgggttc 120
 cgaggatgta ccgtgtggct aacaggtctc tctggtgctg gaaaaacaac gataagttt 180
 gccctggagg agtaccttgt ctcccatgcc atcccttgtt actccctgga tggggacaat 240
 gtccgtcatg gccttaacag aaatctcgga ttctctcctg gggacagaga ggaaaatatc 300
 cgccggattt ctgaggtggc taagctgtt gctgatgctg gtctggctg cattaccagc 360
 tttatattctc cattcgaaa ggatcgtgag aatgcccgc aatacatga atcagcagg 420
 ctgccattct ttgaaatatt tgttagatgca cctctaaata tttgtgaaag cagagacgta 480
 aaaggcctct ataaaagggc cagagctggg gagattaaag gatttacagg tattgattct 540
 gattatgaga aacctgaaac tcctgagcgt gtgcttaaaa ccaattgtc cacagtgagt 600
 gactgtgtcc accaggttgt ggaacttctg caagagcaga acattgtacc ctatactata 660
 atcaaagata tccacgaact ctttgtgccg gaaaacaaac ttgaccacgt ccgagctgag 720
 gctgaaactc tcccttcatt atcaattact aagctggatc tccagtggtt ccaggtttt 780
 agcgaaggct gggccactcc cctcaaaggt ttcatgcggg agaaggagta cttacaggtt 840
 atgcactttg acaccctgct agatgatggc gtgatcaaca tgagcatccc cattgtactg 900

cccgtctctg cagaggataa gacacggctg gaagggtgc gcaagttgt cctggcacat 960
 ggtggacgga gggtagctat cttacgagac gctgaattct atgaacacag aaaagaggaa 1020
 cgctgttccc gtgttgggg gacaacatgt acaaaaacacc cccatatcaa aatggtgatg 1080
 gaaagtgggg actggcttgt tggtgagac cttcaggtgc tggagaaaat aagatggaat 1140
 gatgggctgg accaataccg tctgacacct ctggagctca aacagaaatg taaagaaatg 1200
 aatgctgatg cgggtttgc attccagttg cgcaatcctg tccacaatgg ccatgccctg 1260
 ttgatgcagg acacctgccc caggctcta gagagggct acaagcaccc ggtcctccta 1320
 ctacaccctc tggcggctg gaccaaggat gacgatgtgc ctctagactg gcggatgaaag 1380
 cagcacgcgg ctgtgctcg ggaagggtc ctggatccca agtcaaccat tggtgccatc 1440
 ttccgtctc ccatgttata tgctggcccc acagaggctc agtggactg caggtcccgg 1500
 atgattgcgg gtgccaattt ctacattgtg gggagggacc ctgcaggaat gccccatcct 1560
 gaaaccaaga aggatctgta tgaacccact catggggca aggtctttag catggccct 1620
 ggcctcacct ctgtggaaat cattccattc cgagtggctg cctacaacaa agccaaaaaaa 1680
 gccatggact tctatgatcc agcaaggcac aatgagtttgc acttcatctc aggaactcga 1740
 atgaggaagc tcgcccggga aggagagaat cccccagatg gcttcatggc ccccaaagca 1800
 tggaaggtcc tgacagatta ttacaggtcc ctggagaaga actaa 1845

<210> 10

<211> 1851

<212> DNA

<213> Mus musculus

<400> 10

atgtctgcaa atttcaaaat gaaccataaa agagaccagc aaaaatccac caatgtggc 60
 taccaggccc atcatgttag caggaacaag agaggacaag tggttggAAC caggggagga 120
 ttccgaggat gtaccgttg gctaacaggt ctctctggc ctggaaaaac aaccataagc 180
 tttgcttgg aagagtacct tttatctcac gccatcccat gttactccct ggatggggac 240
 aatgtccgtc atggccttaa taagaacctg ggattctctg ccggggaccg agaagagaat 300
 atccgcccgg a tcgcccgggat ggc当地agctc tttgccgacg ccggcctggc ttgc当地tacc 360
 agctttatct ctcttttg c aaaggatcgt gagaatgccc gaaaaatcca cgaatcagca 420
 ggactcccg tctttgagat cttttagat ggc当地ttttaa atatctgtg aagccgagac 480
 gtaaaaggac tctacaaaacg agcccgagca ggagagatta aagggtttac aggcatcgat 540
 tctgactatg agaaacctgaa aactccagag tttgtgctg agaccaactt gtcttcagta 600
 agcgactgtg tgcaacaggat ggtggactt ttgcaggagc agaacattgt accccacacc 660
 accatcaaag gcatccacga actcttttg cccagaaaaca aagtcgatca aatccgagct 720
 gaggcagaga ctctccatc actaccaatt accaagctgg atctgcagtg ggtgcagatt 780
 ctgagtgaa gctggggccac tcccctcaaa ggcttatgc gggagaagga atacttgcaa 840
 actctacact tcgacactct actggacgat ggagtc当地ca acatgagttt tcccatttgta 900
 ttgcccgtt ctgcccggatgaa caaggcacgg ctc当地agggt gcagcaaatt tgc当地tggatg 960
 tacgaaggc gggactggc tctattacag gaccctgaaat tctatgagca taggaaagag 1020
 gagcgttgg tctcggttg gggaaacagcc actgcaaaagc acccccatat caaaatggc 1080
 atggaaaatgt gggactggct tttgggtgg gacctacagg tgcttagagag aataagggtgg 1140
 gacgatgggc tggaccaata ccccttaacg cctctggaaac tcaaacaagaa gtgtaaagac 1200
 atgaatgctg atggcgttt tgattccag ttgc当地ca ctgtccacaa tggcatgccc 1260
 ctcctgtatgc aggacaccgg cccaggctc ctggagaggg gttacaagca cccagtcctc 1320
 ctgctccacc ctcttggggg ctggaccaag gacgatgacg tacctctggc atggaggatg 1380
 aaacagcatg cagctgtact ggaggaaagg gtc当地ggatc ccaagtcaac tattttgccc 1440
 atctttccat ctctatgtt atacgcttgtt cccacagagg tccagtgca ttgc当地atgc 1500
 cggatgattt caggagccaa tttctacatt gtgggttaggg atcccgagg aatgccccat 1560
 cctgagacaa agaaagaccc atatgaaacc acccacgggg gcaaggctt gatgtggcc 1620
 cctggcctta cctctgtgaa aataattccg ttccgaggatg ctgc当地acaa taaaattaaa 1680
 aaggccatgg acttttatgaa tccagcaagg cacgaggatg ttgacttcat ctc当地act 1740
 cgcatgagga agctcgcccg ggaaggagaa gatccccag atggcttcat ggccccgaaa 1800
 gcgtggaaag tggatgacaga ttactacagg tctctggaga agaccaacta g 1851

<210> 11
<211> 21
<212> DNA
<213> Homo sapiens

<400> 11
gccagccagc atgtcgaaaa t 21

<210> 12
<211> 24
<212> DNA
<213> Homo sapiens

<400> 12
acctgaaaact cctgagcggt tgct 24

<210> 13
<211> 21
<212> DNA
<213> Homo sapiens

<400> 13
gatgtgcctc tagactggcg g 21

<210> 14
<211> 24
<212> DNA
<213> Homo sapiens

<400> 14
gagcacttca gaaagaaaact ctgg 24

<210> 15
<211> 21
<212> DNA
<213> Homo sapiens

<400> 15
catccgccag tctagaggca c 21

<210> 16
<211> 21
<212> DNA
<213> Homo sapiens

<400> 16
agggtgcaga cggtattggc c 21

<210> 17
<211> 23
<212> DNA
<213> Homo sapiens

<400> 17
gtcactcact gtggacaaat tgg 23

<210> 18	
<211> 21	
<212> DNA	
<213> Homo sapiens	
<400> 18	
cacccatcgca atccggcgga t	21
<210> 19	
<211> 20	
<212> DNA	
<213> Mus musculus	
<400> 19	
tctggcacaa agagttcgtg	20
<210> 20	
<211> 22	
<212> DNA	
<213> Mus musculus	
<400> 20	
gccagtttgt aaccgagtat tc	22
<210> 21	
<211> 22	
<212> DNA	
<213> Mus musculus	
<400> 21	
gcaattggat acagagcagc ta	22
<210> 22	
<211> 22	
<212> DNA	
<213> Mus musculus	
<400> 22	
gacaatgtcc gtcatggcct ta	22
<210> 23	
<211> 21	
<212> DNA	
<213> Mus musculus	
<400> 23	
attcccatgg tattgcccgt t	21
<210> 24	
<211> 21	
<212> DNA	
<213> Mus musculus	
<400> 24	

aacgggcaat acaatggaa t	21
<210> 25	
<211> 22	
<212> DNA	
<213> Mus musculus	
<400> 25	
gataaagctg gtgatgcaaa cc	22
<210> 26	
<211> 20	
<212> DNA	
<213> Mus musculus	
<400> 26	
catgggatgg cgtgagatac	20
<210> 27	
<211> 23	
<212> DNA	
<213> Mus musculus	
<400> 27	
cataagctt gctttggaag agt	23
<210> 28	
<211> 21	
<212> DNA	
<213> Homo sapiens	
<400> 28	
gcatgtccag acagacacca c	21
<210> 29	
<211> 333	
<212> DNA	
<213> Homo sapiens	
<220>	
<223> D19Mit13 locus and flanking sequences	
<221> misc_feature	
<222> (1)...(333)	
<223> n = A,T,C or G; at nucleotide positions 23 and 305	
<400> 29	
ctgactatga gaaaacctgaa acnccagagt gtgtgctgaa gaccaacctg tcttcagtaa 60	
gcgactgtgt gcaacagggtg gtggacttt tgcaggagca ggttaggaggg tggttcttgc 120	
cagtgtgttc agtgtgtgtg tgtgtgtgtg tgtgtgtgtg tgcgtgtgca tgtgtgtgtg 180	
catgtgtgtg tgcgtgtgca tgtgtgtgtg ttgaaagata atctgagttt ctttattccc 240	
tggccaatct cagtaactat tgccaatttc gtttcccaca gaacattgta ccccacacca 300	
ccatnaaagg catccacgaa ctctttgtgc cag	333
<210> 30	

<211> 18
<212> DNA
<213> Homo sapiens

<220>
<223> Nucleotide positions +1414 through +1431 of PAPSS2 coding sequence

<400> 30
gatcccaagt caaccatt

18

<210> 31
<211> 6
<212> PRT
<213> Homo sapiens

<220>
<223> Partial PAPSS2 peptide sequence; amino acid residues 472 through 477

<400> 31
Asp Pro Lys Ser Thr Ile
1 5

<210> 32
<211> 18
<212> DNA
<213> Homo sapiens

<220>
<221> mutation
<222> (0)...(0)
<223> Nucleotide positions +1414 through +1431 of PAPSS2 coding sequence with mutation c to a at nucleotide position +1424

<400> 32
gatcccaagt aaaccatt

18

<210> 33
<211> 3
<212> PRT
<213> Homo sapiens

<220>
<223> Partial truncated PAPSS2 peptide sequence; amino acid residues 472-474 plus stop at position 475

<400> 33
Asp Pro Lys
1